

Knowledge and attitude towards hemoglobinopathies among Saudi population: A Cross sectional study in Riyadh, Saudi Arabia

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ABSTRACT

Background: Hemoglobinopathies are caused by genetic variations resulting in either a qualitative or quantitative abnormality in Hemoglobin. These diseases are prevalent in Kingdom of Saudi Arabia due to consanguineous marriages, which happens to be traditional practice in Saudi Arabia, as a result, faulty genes associated with hemoglobinopathies if present, get transmitted from parents to their offsprings. **Methodology:** In this study, we have measured the level of knowledge and attitude of Riyadh population towards hemoglobinopathies specifically thalassemia and sickle cell anemia. This is a cross-sectional study which was conducted between December 2020 and May 2021 in Riyadh city of Saudi Arabia with the help of self-administered questionnaire. The sample of the study consisted of 486 individuals from Riyadh city, most of whom were Saudi nationals. **Results:** Among all study participants, 42.4% had consanguineous marriages between their parents; 67.1% had some knowledge about hemoglobinopathies; 43.6% had some knowledge of thalassemia; 74.5% had some knowledge of sickle cell anemia; 34.4% had awareness about the importance of premarital screening. Regression analysis showed the association of different demographic variables with the awareness of hemoglobinopathies and premarital screening to be statistically significant ($p<0.05$). **Conclusion:** It is concluded from this study that, awareness for importance of premarital screening was very less among study participants, so more awareness is required to minimize the prevalence of haemoglobinopathies among general population of Saudi Arabia.

Keywords: Knowledge, attitude, hemoglobinopathies, premarital screening.

1. INTRODUCTION

Hemoglobinopathies (HGP) are defined as set of clinical diseases that result from a genetically determined abnormality of the structure or synthesis of the

hemoglobin molecule. They are the most common single gene disorders in the world, majority of cases happen in low-income countries, which can lead to devastating economical results to these countries (McKenzie et al., 2020). HGP can be classified into two groups, quantitative and qualitative. In the qualitative group, the defect happens in the structure of hemoglobin, Sickle Cell Anemia (SCA) is a classic example for this group. On the other hand, the quantitative group, thalassemia is the representative of this group with the defect happening in the rate of hemoglobin synthesis (Chandrakasan and Malik, 2014). The main treatment for these diseases is bone marrow transplantation. However, bone marrow transplantation is not suitable for all cases. Moreover, it is required a lot of lab investigation which is time and money consuming. HGP have high impact on patient social and psychological life and the society (Memish et al., 2011). It is estimated that 330,000 affected births occur annually with SCA (83%), and thalassemia (17%). WHO reported in 2008 that 5.2% of the world population are carriers of a significant HGP causing genomic variant, and that 24% are carriers of all known HGP causing genomic variant that are both pathogenic and nonpathogenic forms (Goonasekera et al., 2018).

Endogamy marriages (consanguineous marriages) in Arab and gulf country are high and that's for many traditional and cultural reasons (Gosadi, 2019). Saudi Arabia initiated a program called premarital screening (PMS) and genetic counseling program (PMSGC) in 2004 the goal of the program is to reduce the risk of genetic and infectious diseases to the couple and offspring (El Mouzan et al., 2008).

The aim of this study is to explore different levels of knowledge and attitude towards the role of heredity in HGP specifically thalassemia and SCA among Saudi population. Additionally, this study also investigates the awareness for the importance of PMS programs among Saudi population to control the HGP in Saudi Arabia.

2. METHODOLOGY

A Cross-sectional study was conducted between December 2020 and May 2021 in Riyadh city of Saudi Arabia. A self-administered questionnaire was distributed electronically to a total of 486 eligible participants to measure the knowledge and attitude of general population towards HGP and PMS program (PMS). The questions were gathered specifically from several previously published and validated surveys to measure the level of the knowledge and attitude towards thalassemia, SCA, and quality of life for HGP patients. Questions were designed, pooled, validated, and selected by an experienced public health practitioner. Ethical approval was obtained from the Institutional Review Board of Al-Imam Muhammad Ibn Saud Islamic University in Riyadh, Saudi Arabia. To collect data, the self-administered questionnaire was sent through social media (twitter, whats app etc). The sampling technique used was random sampling. The questions were translated from English to Arabic by a bilingual translator whose mother tongue is Arabic. All participants were informed about the study, objectives, and consent was obtained.

Only completed questionnaires with informed consent from the participants were considered in the analysis. The inclusion criteria for the study were adult Riyadh residents who can speak Arabic or English. Non-Riyadh residents, non-Arabic language speakers, minor ages (less than 18 years) were excluded. The target sample was 385 using the online Survey software (<http://www.raosoft.com/samplesize.html>) at Confidence Level was 95%, margin of error was 5%. The questionnaire was composed of three parts:

Sociodemographic characteristics:

This section elicited sociodemographic information of participants and there were five questions asking about age, gender, marital status, education, and area of residence in Riyadh.

Knowledge about thalassemia and SCA

The questions in this section of the questionnaire aimed to assess the knowledge level about thalassemia, SCA and to explore the attitude towards HGP patients in Riyadh. We asked general questions about thalassemia such as inheritance, transmission, treatability, symptoms of thalassemia, effects on daily life, the need for blood transfusion, and source of information. Other questions have been asked about their attitude towards HGP patients such as asking if they think HGP patients could work like everyone else or should be supported by the government.

Knowledge and attitude towards PMS

The questions in this section of the questionnaire aimed to assess knowledge and attitude towards PMS among the general population in Riyadh. It contained 6 statements ranging from strongly agree to strongly disagree. We asked the participants if they think PMS is a preventive measure or not. Other questions have been asked to evaluate their willingness to carry out PMS.

Statistical analysis

Descriptive statistics have been generated to describe the demographic variables of study participants. Frequencies and percentages have been calculated for all the categorical parameters. Means and standard deviations were calculated for the continuous variable of age. Regression analysis was performed to investigate the association of the independent variables of age, sex, marital status and education with dependent variables of knowledge of thalassemia, knowledge of SCA, and awareness towards importance of premarital screening. The criterion for significance was set at $p < 0.05$. All data analysis was done by using IBM SPSS statistics version 20.

3. RESULTS

Table 1 show the demographic characteristics of the study participants which consisted of 486 individuals from Riyadh, most of whom were Saudis. Most of the participants were females (64.8%). Their ages ranged from 14 to 70 years, with mean age of males was 30.75 years and mean age of females was 36.91 years. Most of the participants (96.9%) were Saudi nationals.

Table 1 The demographic characteristics of the study participants

Variable	Group	Frequency	Percent
Sex	Female	315	64.8%
	Male	171	35.2%
Age	Female (Mean \pm SD)	36.91 \pm 13.62	-
	Male (Mean \pm SD)	30.75 \pm 12.74	-
Marital status	Married	250	51.4%
	Single	236	48.6%
Education	Doctorate	25	5.1%
	Master	36	7.4%
	Bachelor	321	66.0%
	Secondary school	95	19.5%
	Less than secondary	9	1.9%
Area of residence in Riyadh	South of Riyadh	37	7.6%
	East of Riyadh	194	39.9%
	North of Riyadh	139	28.6%
	West of Riyadh	55	11.3%
	Center of Riyadh	61	12.6%
Nationality	Saudi	471	96.9%
	Non-Saudi	15	3.1%

Table 2 shows the questions related to thalassemia. The participants who had heard about HGP were 67.1% of the total study participants; these participants represented a little more than two-thirds of the study participants. The participants who had consanguineous marriage between parents were 42.4%. The participants who had heard about thalassemia were 43.6%. The participants, who had heard about thalassemia, indicated that they had heard about thalassemia from social media or Internet. About 61.8% of participants who had heard about thalassemia were certain that the patient with thalassemia requires blood transfusion. While 14.6% of who had heard about thalassemia responded that the patient with thalassemia not requires blood transfusion. The rest 23.6% of them did not had knowledge about this question. Among total participants, 54.7% had heard about thalassemia were positive that thalassemia can be treated by blood transfusion. While the participants who believed that

thalassemia can be treated with antibiotics and vaccines were 7.5% and 4.7% respectively. The rest 33% of those who had heard of thalassemia knew nothing about its treatment. The most of those who had heard about thalassemia (95.8%) were certain that if both parents had the thalassemia trait, there would be a chance of having a child with this disease. The participants who believe that there is a cure for thalassemia were 10.8%, while the participants who believe that there was no cure for thalassemia were 45.3%. The rest 43.9% participants did not have knowledge about this question. The participants who believed that individuals who had thalassemia cannot live normal life even with appropriate treatment were 26.9%, while participants who reject this question were 61.3%. The participants who believed that a person with thalassemia trait (carrier) can be healthy and shows no symptoms of the disease were 65.6%, which was higher than the participants who reject this information (22.6%).

The participants who believe that a person with thalassemia disease has a higher risk of infections and/or illnesses were 73.1%. Most of the respondents who had heard of thalassemia (97.6%) believe that thalassemia was not an infectious disease. Most of the respondents who had heard of thalassemia (94.8%) decided that patient with thalassemia may appear pale or yellow. The participants who decided that a patient with thalassemia may feel excessive fatigue without doing any activities were 88.7%. Most of the respondents who had heard of thalassemia (91.5%) think that patients with thalassemia should be supported by the government or should work like everyone else. Most of the respondents who had heard of thalassemia (94.8%) believe that patients with thalassemia could marry and had a family. The participants who know someone with thalassemia from family/relatives, friends and neighbors were 21.7%, 13.2% and 3.3% respectively. The rest 61.8% of them do not know someone with thalassemia.

Table 2 Frequency distribution of responses related to thalassemia among study participants

S. No.	Question	Responses	Frequency	Percent
1	Have you heard about HGP?	Yes	326	67.1%
		No	160	32.9%
2	Is there a family relationship between parents (Consanguinity)?	Yes	206	42.4%
		No	280	57.6%
3	Have you heard of thalassemia?	Yes	212	43.6%
		No	274	56.4%
4	Have you ever heard about Thalassemia from any of these sources?	Books	3	1.4%
		University, college or school	7	3.3%
		Education (studying)	12	5.7%
		Hospital	8	3.8%
		Social media or Internet	107	50.5%
		Newspapers	17	8.0%
		Family or friends	53	25.0%
		TV	5	2.4%
5	Patient with thalassemia requires blood transfusion?	Yes	131	61.8%
		No	31	14.6%
		I don't know	50	23.6%
6	Thalassemia can be treated by?	Blood transfusion	116	54.7%
		Vaccines	10	4.7%
		Antibiotics	16	7.5%
		I don't know	70	33.0%

S. No.	Question	Responses	Frequency	Percent
7	If both parents have the Thalassemia trait, there is a chance of having a child with Thalassemia disease?	Yes	203	95.8%
		No	9	4.2%
8	Thalassemia is a disease that runs in the family?	Yes	199	93.9%
		No	13	6.1%
9	There is a cure for thalassemia?	Yes	23	10.8%
		No	96	45.3%
		I don't know	93	43.9%
10	Individuals who have thalassemia cannot live normal lives even with appropriate treatment.	Yes	57	26.9%
		No	130	61.3%
		I don't know	25	11.8%
11	A person with thalassemia trait (carrier) is otherwise healthy and shows no symptoms of the disease.	Yes	139	65.6%
		No	48	22.6%
		I don't know	25	11.8%
12	A person with thalassemia disease has a higher risk of infections and/or illnesses.	Yes	155	73.1%
		No	11	5.2%
		I don't know	46	21.7%
13	Do you believe thalassemia is not an infectious disease?	Yes	207	97.6%
		No	5	2.4%
14	Patient with thalassemia may appear pale or yellow?	Yes	201	94.8%
		No	11	5.2%
15	Patient with thalassemia may feel excessive fatigue without doing any activities.	Yes	188	88.7%
		No	17	8.0%
		I don't know	7	3.3%
16	Do you think patients with thalassemia should be supported by the Government or should work like everyone else?	Yes	194	91.5%
		No	18	8.5%
17	Do you believe patients with thalassemia could marry and have a family?	Yes	201	94.8%
		No	11	5.2%
18	Do you know someone with thalassemia?	Family or relatives	46	21.7%
		Neighbors	7	3.3%
		Friends	28	13.2%
		I don't know	131	61.8%

Table 3 shows questions related to SCA. The participants of those participants who had heard about SCA were 74.5% of the total study participants. Highest participants who had heard about SCA (66%) decided that SCA was a disease that runs in the family, while the 12.4% of them reject this information. The rest participants (21.5%) of them not had knowledge about this matter. Most of those who heard about SCA (81.5%) decided that if both parents had the sickle cell trait, there was a chance of having a child with SCA. Participants of 50.6% of total participants, who had heard about SCA, responded that the patient with SCA requires blood transfusion. Most of the respondents who had heard of SCA (80.9%) believe that SCA is not an infectious disease. Participants who had responded that patient with SCA may appear pale or yellow were 76.8%. The participants who had responded that a patient with SCA may feel excessive fatigue without doing any activities were 65.5%. The majority of the respondents who had heard of SCA (66.3%) think that patients with SCA should be supported by the government or should work like everyone else. Most of the respondents who had heard about SCA (73.2%) believe that patients with SCA could marry and had a family. Participants of 38.4% among those participants who had heard about SCA responded that the SCA can be treated by blood transfusion while 48.9% of those who had heard about SCA knew nothing about its treatment.

Table 3 Frequency distribution of responses related to sickle cell anemia among study participants

S. No.	Question	Responses	Frequency	Percent
1	Have you heard of sickle cell anemia?	Yes	362	74.5%
		No	124	25.5%
2	Sickle cell is a disease that runs in the family.	Yes	239	66.0%
		No	45	12.4%
		I don't know	78	21.5%
3	If both parents have the sickle cell trait, there is a chance of having a child with sickle cell disease?	Yes	295	81.5%
		No	20	5.5%
		I don't know	47	13.0%
4	Patient with sickle cell disease requires blood transfusion?	Yes	183	50.6%
		No	49	13.5%
		I don't know	130	35.9%
5	Do you believe sickle cell disease is not an infectious disease?	Yes	293	80.9%
		No	29	8.0%
		I don't know	40	11.0%
6	Patient with sickle cell disease may appear pale or yellow?	Yes	278	76.8%
		No	16	4.4%
		I don't know	68	18.8%
7	Patient with sickle cell disease may feel excessive fatigue without doing any activities?	Yes	237	65.5%
		No	20	5.5%
		I don't know	105	29.0%
8	Do you think patients with sickle cell disease should be supported by the government or should work like everyone else?	Yes	240	66.3%
		No	30	8.3%
		I don't know	92	25.4%
9	Do you believe patients with sickle cell disease could marry and have a family?	Yes	265	73.2%
		No	21	5.8%
		I don't know	76	21.0%
10	Sickle cell disease can be treated by?	Blood transfusion	139	38.4%
		Vaccines	19	5.2%
		Antibiotics	27	7.5%
		I don't know	177	48.9%

Figure 1 - 8 show responses related to PMS examination, which reveals that 42.2% of the total study participants strongly disagree about going through marriage when the risk of a hereditary disease (figure 1). Most of the respondents (83.1%) of the total study participants were strongly agreed about PMS were a preventive measure (figure 2). Most of the respondents (84.2%) were

strongly agree about PMS must be mandatory by law (figure 3). Many of the respondents (78.6%) were strongly agree about they would carry out PMS even if it was not mandatory (figure 4). About half (49.8%) of the total study participants size were strongly agree about they would demand implementation of a law that prohibits incompatible marriages (figure 5). More than half of the total study participants (54.5%) were strongly agreed about they would cancel the marriage with incompatible results (figure 6). However, only 5.9% had responded not to cancel the marriage with incompatible results. Participants who were agreeing about they prefer consanguineous marriage were 59%, while the 41% participants did not agree to prefer consanguineous marriage (figure 7). Most of the respondents (86%) agree to carry out PMS for the reason of preventing disease transmission to offspring, while the 6.2% participants prefer PMS for the reason of ensuring health of the partner, participants of 5.1% for the reason of preventing disease transmission to themselves, the participants who prefer pre-marital examination because it was legally required were 2.7% (figure 8).

Frequency distribution of responses related to premarital screening among study participants

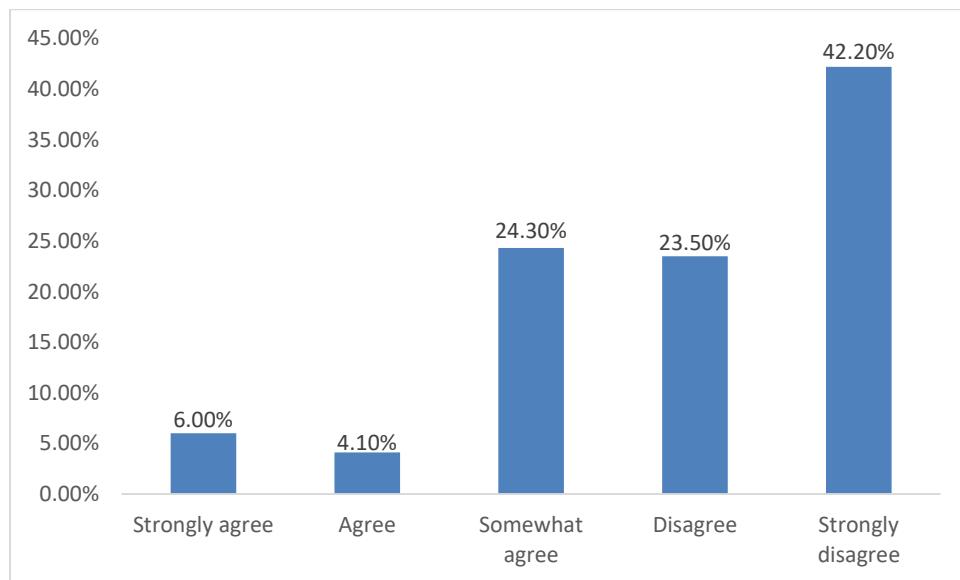


Figure 1 Would you go through marriage even with the risk of a hereditary disease?

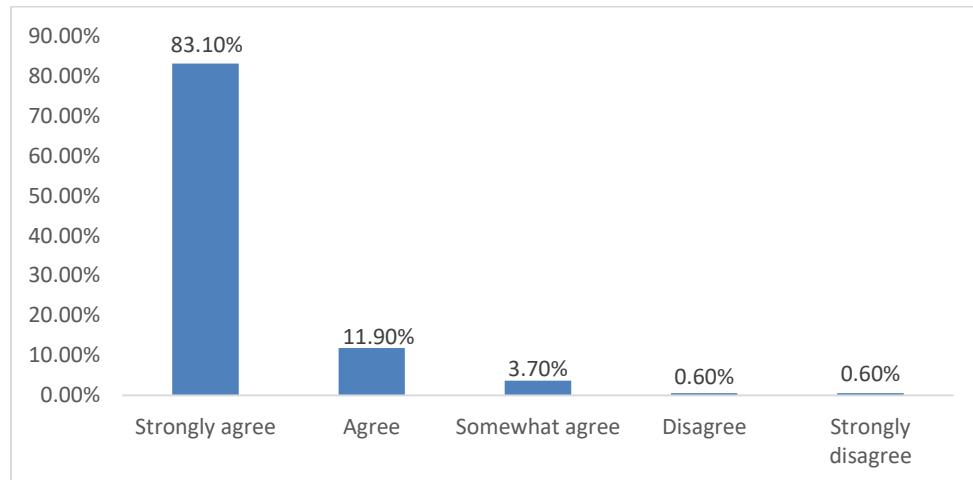


Figure 2 Do you think PMS is a preventive measure?

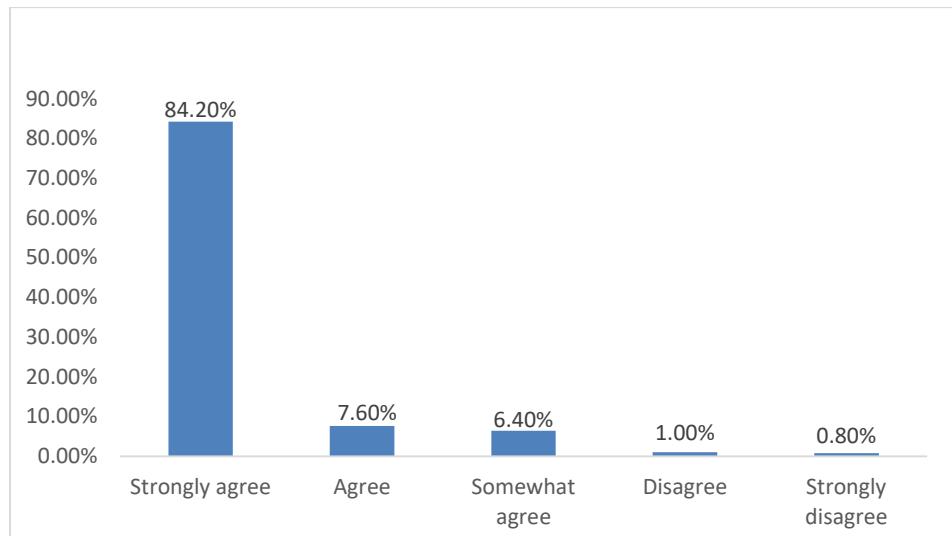


Figure 3 Do you think PMS must be mandatory by law?

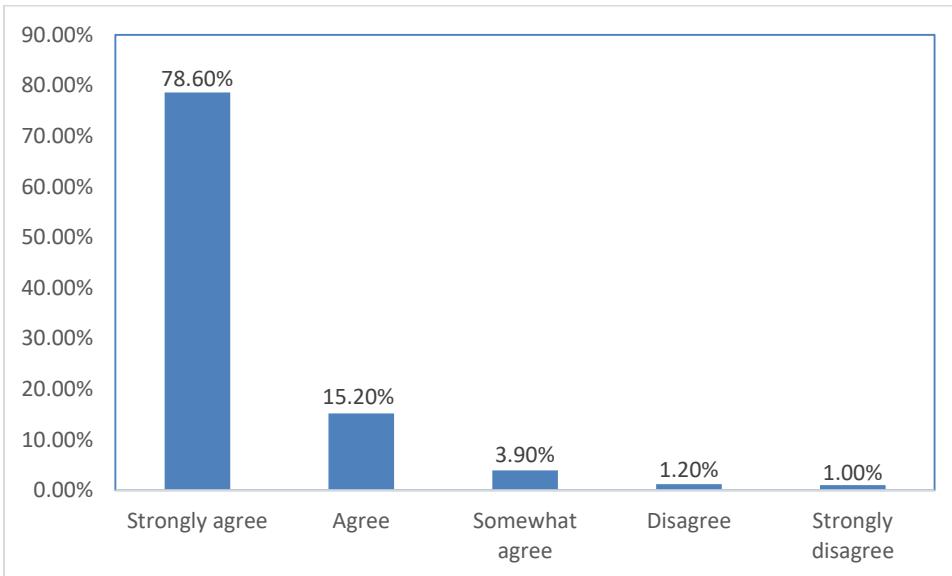


Figure 4 Would you carry out PMS even if it is not mandatory?

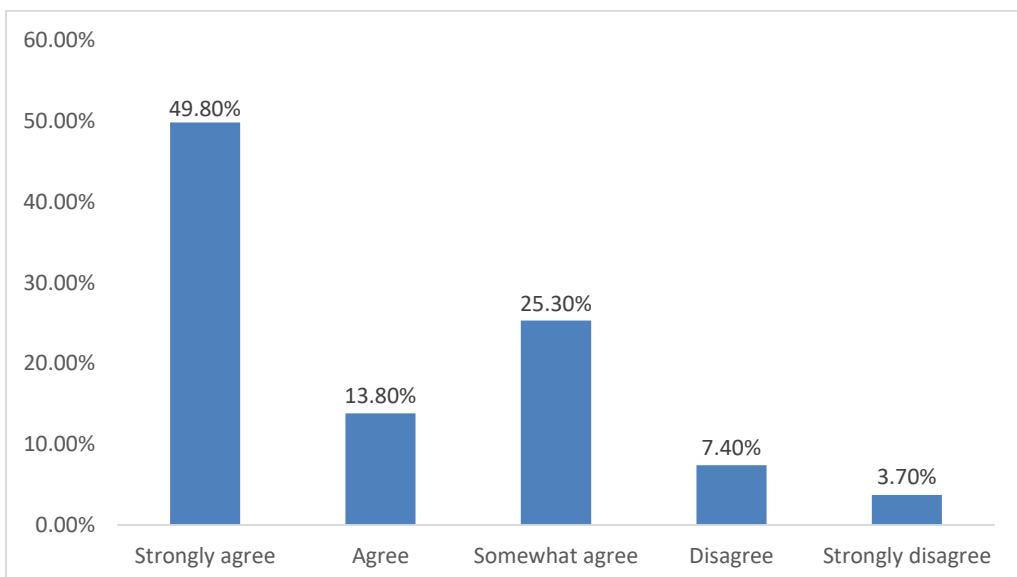


Figure 5 Would you demand implementation of a law that prohibits incompatible marriages?

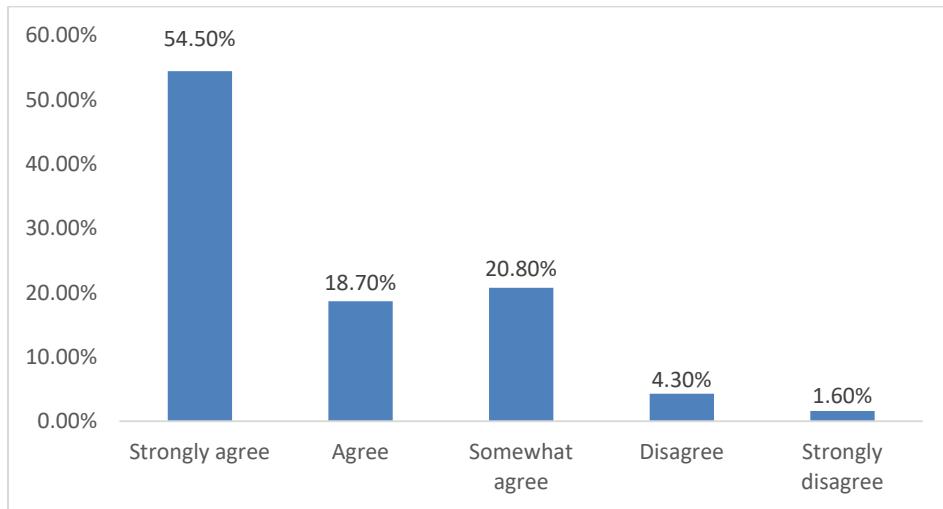


Figure 6 Would you cancel the marriage with incompatible results?

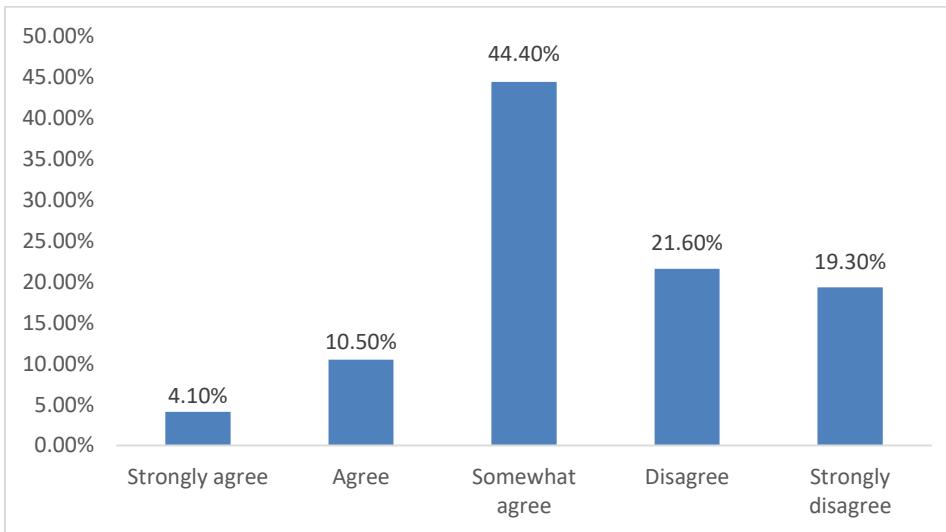


Figure 7 Would you prefer consanguineous marriage?

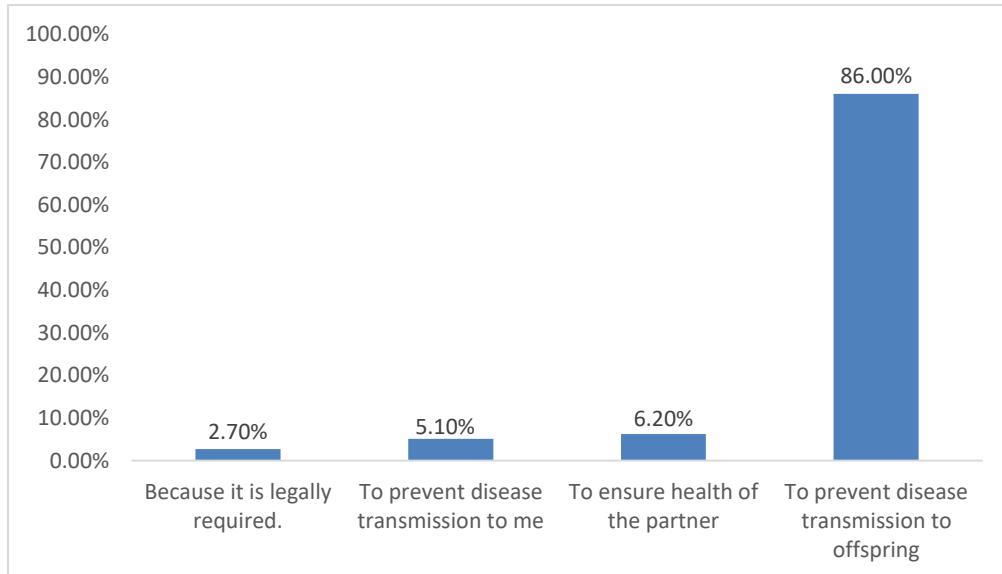


Figure 8 You would carry out PMS for which of the following motives?

Table 5 shows the regression analysis focused on three domains of study; knowledge of thalassemia, knowledge of sickle cell, and awareness towards importance of premarital screening. On the other hand, the independent variables that were regressed include age, sex, social status, and education. In this respect, 3 regression output were produced as detailed in the table (5). For knowledge of thalassemia, the results were statistically highly significant ($r = .299$, $p = .000$), for knowledge of SCA, the results were also statistically highly significant ($r = .394$, $p = .000$) and significant for premarital sex ($r = .155$, $p = .022$). In this regard, it can be implicated that to a small extent, the variables age, sex, marital status and education can be used to predict the knowledge of thalassemia and SCA and also for premarital examination. However, the r values were relatively small and hence show a weak predictability of knowledge of thalassemia, SCA and premarital examination using the study variables.

Table 5 Regression analysis for comparison of different study variables with knowledge of thalassemia, knowledge of sickle cell, and premarital screen

Interest variables	Coefficients	R value	P value
Knowledge of thalassemia		.299	0.00
Age	-.007		
Sex	-.358		
Marital status	-.241		
Education	.001		
Knowledge of sickle cell anemia		.394	0.00
Age	-.001		
sex	-.314		
Marital status	-.046		
Education	.015		
Awareness towards importance of premarital screening		.155	0.022
Age	.002		
sex	-.062		
Marital status	.093		
Education	-.030		

4. DISCUSSION

HGP are autosomal recessive disorders, caused by a genetic mutation affecting the hemoglobin moiety, leading to either a quantitative (thalassemia) or qualitative SCA. Considering that HGP are quite prevalent in the kingdom of Saudi Arabia we suspected a low level of public awareness and basic knowledge about them, which needed to be quantified and measured the deficits in knowledge of general Saudi population of Riyadh city. Awareness for the risk factors of HGP is weak among Saudi population, which is documented in a cross sectional study by El-Mouzan (2008), who investigated the prevalence of consanguinity to be 56% with the first-degree cousin (33.6%) being more common than all others (22.4%) and that can lead to transmission of many genetic disorders (Alsaeed et al., 2017). The prevalence rate in Saudi Arabia for β -thalassemia major increased from 1 in 2011 to 1.6 in 2015, which suggests that increased number of cases and the persistence of consanguinity is an indicator for the need of higher education and awareness programs for HGP (Ayman et al., 2008).

In the present study, data showed that there is a great disparity between participants who knew about thalassemia (43.6%) and those who knew about SCA (74.5%), this indicates the need for increased public health efforts in raising awareness about HGP, moreover basic knowledge about management of HGP was decreased compared to the knowledge of genetic role in transmission of HGP. Our study showed some agreement with the findings of a previous study conducted in Saudi Arabia by Ayman et al., (2008) in which researchers reported that 80% of the study participants were aware about the role of inheritance in HGP and 60% were agreed with the importance of PMS. Looking at the national prevalence of thalassemia and SCA in Saudi Arabia, high number of cases equals high priority to educate people about these diseases, in order to have a better response to preventive measures and raise the awareness about HGP; also, considering the current mainstay treatment consisting of blood transfusions which can lead to a lot of complications due to iron overload, leading to tissue damage and Iron toxicity and death, another feasible option is allogenic

Hematopoietic Stem Cell Transplant (HSCT), nevertheless, most patients don't have a Human Leucocyte matched sibling (Chandrakasan and Malik, 2014).

One of the earliest findings of the PMS program for the period between 2004 and 2005 in Saudi Arabia was published by Alhamdan et al., (2007). During the starting of program, itwas observed that counselling had limited benefit in reducing marriage of risky couples, where about 90% of risky couples proceeded with their marriage despite informing them about the probability of having offspring affected with HGP. Memish and Saeedi (2011) reported that, the outcomes of PMS covering a longer period between 2004 and 2009, a marked reduction in overall prevalence of thalaseemia traits was observed when compared to the study of Alhamdan et al., (2007).

Another related factor enhancing the spread of HGP in Saudi Arabia is the prevalent consanguinity in Saudi communities, which happens to be a common tribal tradition, leading to transmitting of these faulty genes to their offspring and so on (Memish and Saeedi, 2011). The aim of these PMS and genetic counseling programs is to reduce the prevalence of HGP by preventing at-risk marriages and performing PND followed by termination of the affected fetuses, which is offered in some countries (Saffia and Howard, 2015). These are the reasons to be evident for the importance of measuring public knowledge of HGP. PMS is a very useful tool in primary prevention and is the only primary preventative measure in diseases such as HGP; there were good indicators of raised awareness about the importance of PMS in the detection and prevention of hereditary diseases. The majority of our study participants strongly agreed that PMS should be mandatory by law (84.2%) another good indication of the awareness of Primary prevention.

5. CONCLUSION

In conclusion, present study reports that there is less awareness about the hereditary role in the development of HGP and less awareness about the importance of PMS programs in general population of Riyadh city. HGP are prevalent in Saudi Arabia, so studying them in all their aspects is crucial for community health and preventive medicine. It is recommended that, more public health education and awareness about HGP and PMS should be executed in general population of Riyadh city. Low sample size is the limitation of this study, so such kind of more future researches with large sample size are suggested to paint a clear picture of deficits and requirements in awareness and knowledge among Saudi masses for planning of better effective strategies to minimize the prevalence of HGP in Saudi Arabia.

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Ethical approval

The study was approved by the Medical Ethics Committee of Al-Imam Mohammad Ibn Saud Islamic University, ethical approval code: 103-2020 (registration HAPO-01-R-0011).

Ethical approval

The study was approved by the research center of Riyadh Elm University (SRS/2020/14/193/185).

Conflicts of interest

The authors declare that they have no conflict of interest.

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This study has not received any external funding.

Data and materials availability

All data associated with this study are present in the paper.

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